Disease Name — Congenital Hypothyroidism

Acronym	CH
Disease Classification	Endocrine Disorder
Symptom onset	Clinical signs of hypothyroidism often do not appear until the infants is 3-4 months of age, thus it is most likely that affected infants will have already suffered irreversible brain damage before signs of the disease begin to appear. Many times the early diagnosis relies almost solely on the results of the newborn screening.
Symptoms	An affected infant may have prolonged neonatal jaundice, growth failure, lethargy, poor appetite and constipation.
Natural history without treatment	Even mild hypothyroidism can lead to severe mental retardation and growth retardation if untreated. Development is delayed early on, often indicated by failure to meet normal milestones.
Treatment	Daily oral thyroxine medication to prevent problems, treatment must begin shortly after birth and is lifelong.
Emergency Medical Treatment	See sheet from American College of Medical Genetics (attached) or for more information, go to website: http://www.acmg.net/StaticContent/ACT/Primary_TSH.pdf
Inheritance	Although this disorder is detectable at birth, it is not an inherited disorder. Hypothyroidism does not follow any type of pattern as to whom it will affect and randomly affects infants from almost every origin.
General population incidence	Estimated to affect 1 in 4,500 births
OMIM Link	http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=201910
Genetests Link	www.genetests.org
Support Group	MAGIC Foundation for Children's growth (MAGIC) http://www.magicfoundation.org National Organization for Rare Diseases http://www.rarediseases.org

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